



ACAD8 gene

acyl-CoA dehydrogenase family member 8

Normal Function

The *ACAD8* gene provides instructions for making an enzyme called isobutyryl-CoA dehydrogenase (IBD). This enzyme is found in mitochondria, the energy-producing centers inside cells. The IBD enzyme is involved in breaking down proteins from food. Specifically, this enzyme is responsible for the third step in the breakdown of a protein building block (amino acid) called valine. The IBD enzyme converts a molecule called isobutyryl-CoA into a molecule called methylacrylyl-CoA. Other enzymes further break down methylacrylyl-CoA into molecules that cells can use for energy.

Health Conditions Related to Genetic Changes

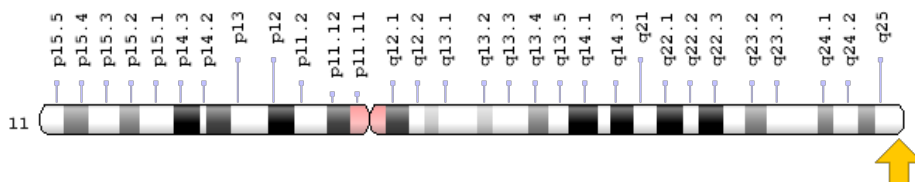
isobutyryl-CoA dehydrogenase deficiency

At least 19 mutations in the *ACAD8* gene have been found to cause isobutyryl-CoA dehydrogenase (IBD) deficiency. Some of these mutations reduce the activity of the IBD enzyme, while other mutations prevent the gene from producing any functional enzyme. As a result, valine is not broken down properly. An inability to process valine may lead to reduced energy production and the features of IBD deficiency.

Chromosomal Location

Cytogenetic Location: 11q25, which is the long (q) arm of chromosome 11 at position 25

Molecular Location: base pairs 134,253,532 to 134,265,858 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ACAD-8
- ACAD8_HUMAN
- Activator-recruited cofactor 42 kDa component
- acyl-CoA dehydrogenase family, member 8
- acyl-coenzyme A dehydrogenase 8
- ARC42
- FLJ22590

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Major pathways of branched-chain amino acid metabolism
<https://www.ncbi.nlm.nih.gov/books/NBK20436/figure/A3097/>

Genetic Testing Registry

- GTR: Genetic tests for ACAD8
<https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=27034%5Bgeneid%5D>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ACAD8%5BTIAB%5D%29+OR+%28ACAD-8%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- ACYL-CoA DEHYDROGENASE FAMILY, MEMBER 8
<http://omim.org/entry/604773>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ACAD8%5Bgene%5D>
- HGNC Gene Family: Acyl-CoA dehydrogenase family
<http://www.genenames.org/cgi-bin/genefamilies/set/974>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=87

- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/27034>
- UniProt
<http://www.uniprot.org/uniprot/Q9UKU7>

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